PARENT FACT SHEET

DISORDER

Beta Ketothiolase (BKT)

CAUSE

BKT occurs when the mitochondrial acetoacetyl-CoA thiolase enzyme (MAT) is either missing or not working properly. This enzyme's job is to help break down the amino acid, isoleucine. Isoleucine is found in all foods that contain protein. When a child with BKT eats food containing isoleucine, harmful substances called organic acids build up in the blood and urine, causing metabolic crisis and brain damage.

IF NOT TREATED

Each child with BKT has slightly different effects. The first symptoms often start around age one, although babies can have symptoms earlier or later than this. BKT causes episodes of illness called metabolic crises, which can cause brain damage, learning disabilities, mental delays, and other problems.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment should begin immediately and will continue throughout life.

- Your child needs to avoid going a long time without food. This is to avoid a metabolic crisis. Children with BKT should not go more than 4 to 6 hours without food and some may require more frequent feedings. Your child's metabolic specialist will advise you as to whether your child needs to eat more often than usual and how to space your child's meals.
- The dietician will help develop a food plan that meets your child's needs. Any changes in diet should only be made by the dietician. While some children with BKT can eat normal amounts of protein, others will need to be on a low-protein diet.
- Periodic urine tests should be done to test the level of ketones. This can be done at home or at the doctor's office. Ketones are substances formed when body fat is broken down to use for energy.
- This happens after going without food for long periods of time, during illness, or during periods of heavy exercise. Ketones in the urine may signal the start of a metabolic crisis.
- Contact your child's doctor immediately at the start of any illness. Children with BKT may require hospitalization to prevent metabolic crisis.

IF TREATED

If treatment is started early and metabolic crises do not occur, your child is likely to have normal growth and intelligence. Even with treatment, some children still have repeated episodes of metabolic crises, which can cause brain damage resulting in learning difficulties, mental delays and other problems.